

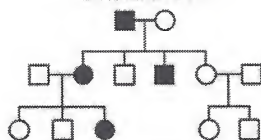
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MALATTIE MUSCOLARI EZIOLOGIA

Forme Geneticamente Determinate

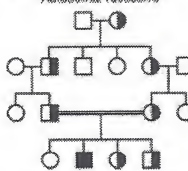
Strutturale	Miopatie Distrofiche Miopatie Congenite
Metabolica	Glicogenosi Lipidosi Metabolismo Glicidico Metabolismo Purinico Miopatie Mitocondriali
Canalopatie	Canali K: Paral Period Ipokaliemica Canali Na: Paral Period Iperkaliemica Canali Cl: Miotonia (non distrofica) Canali Ca: Paral Ipokaliemica Ipertermia Maligna
Miotonia	Congenita Distrofica

A Autosomal dominant



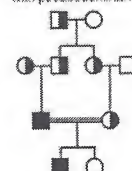
Source: Longo DL, Fauci AS, Kasper DL, Hauser SL, Jameson JL, Loscalzo J: Harrison's Principles of Internal Medicine, 18th Edition; www.accessmedicine.com
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B Autosomal recessive

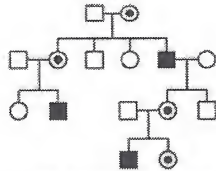


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Autosomal recessive with pseudodominance

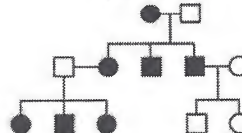


C X-linked

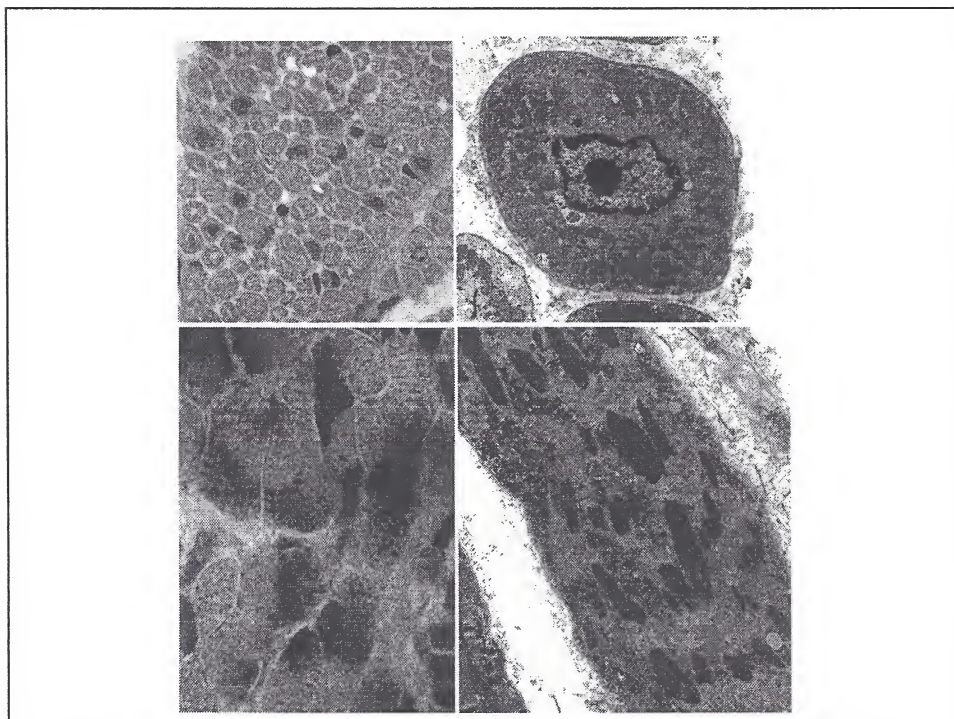
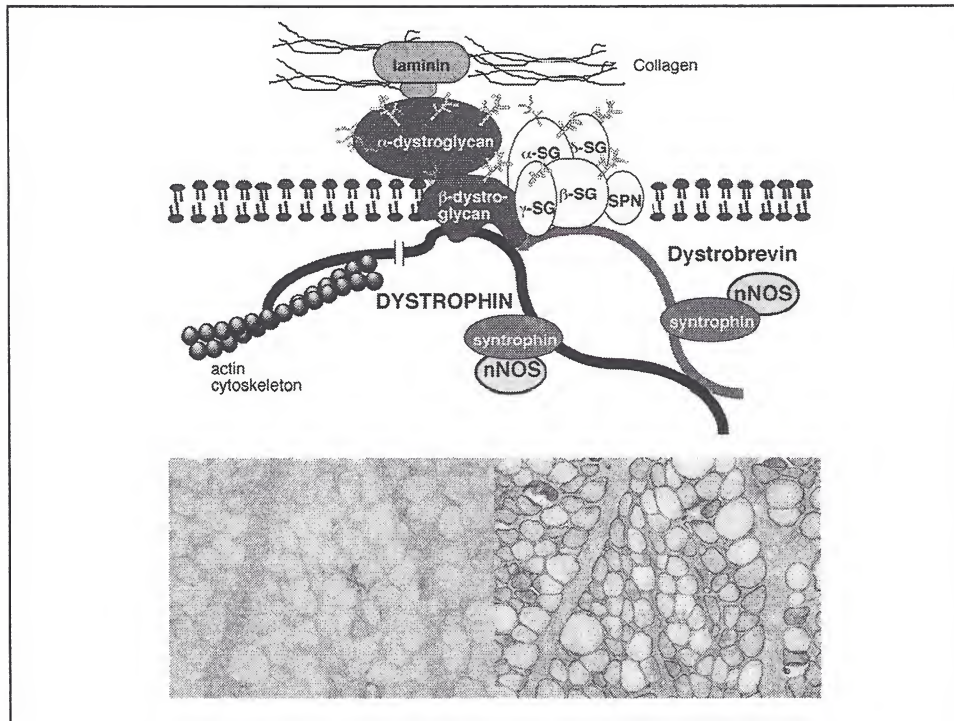


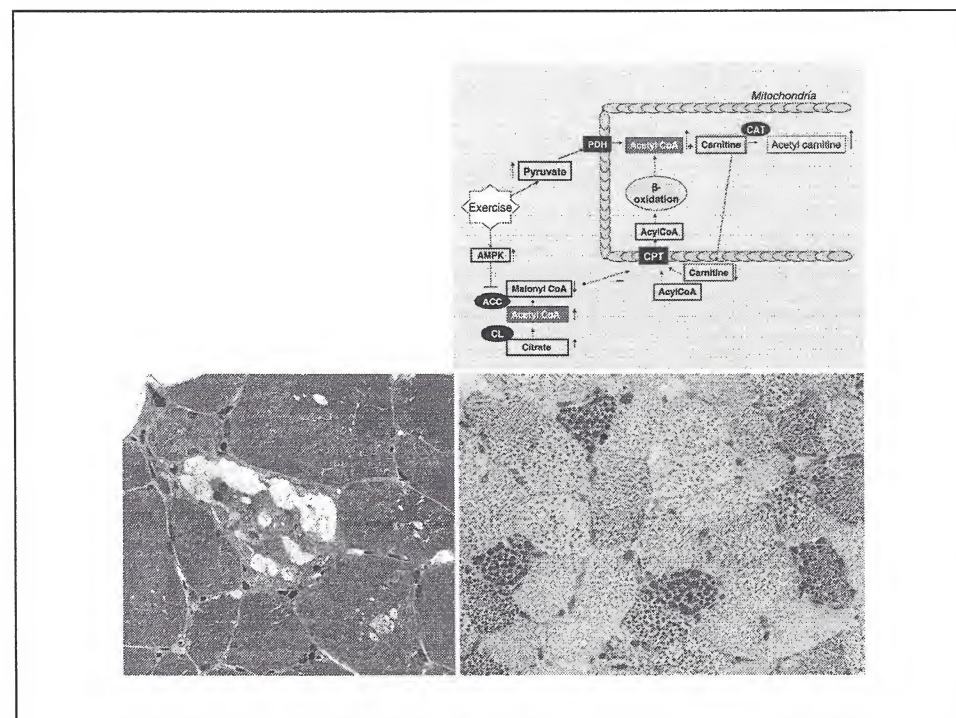
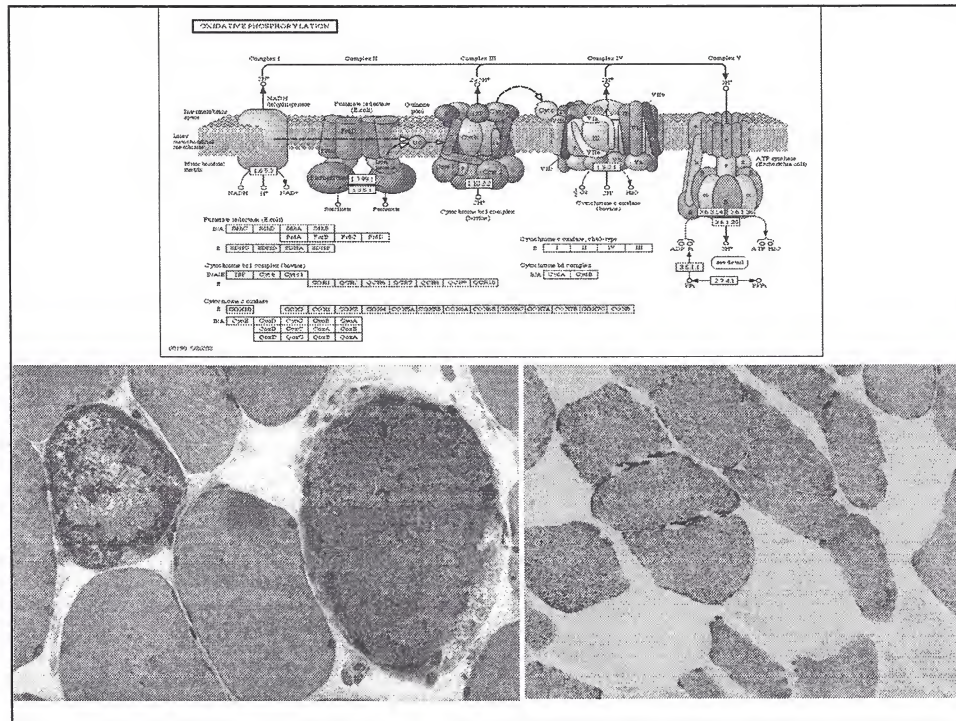
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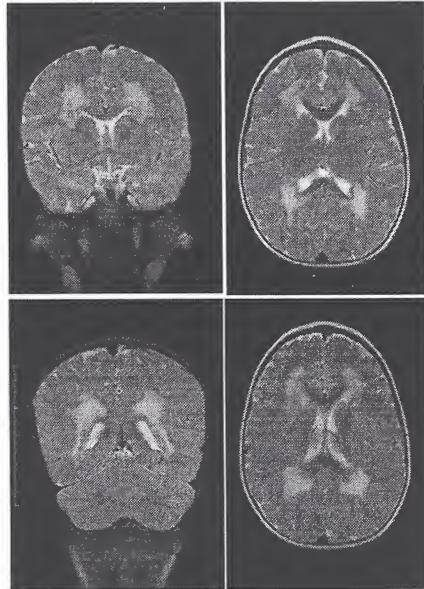
D Mitochondrial



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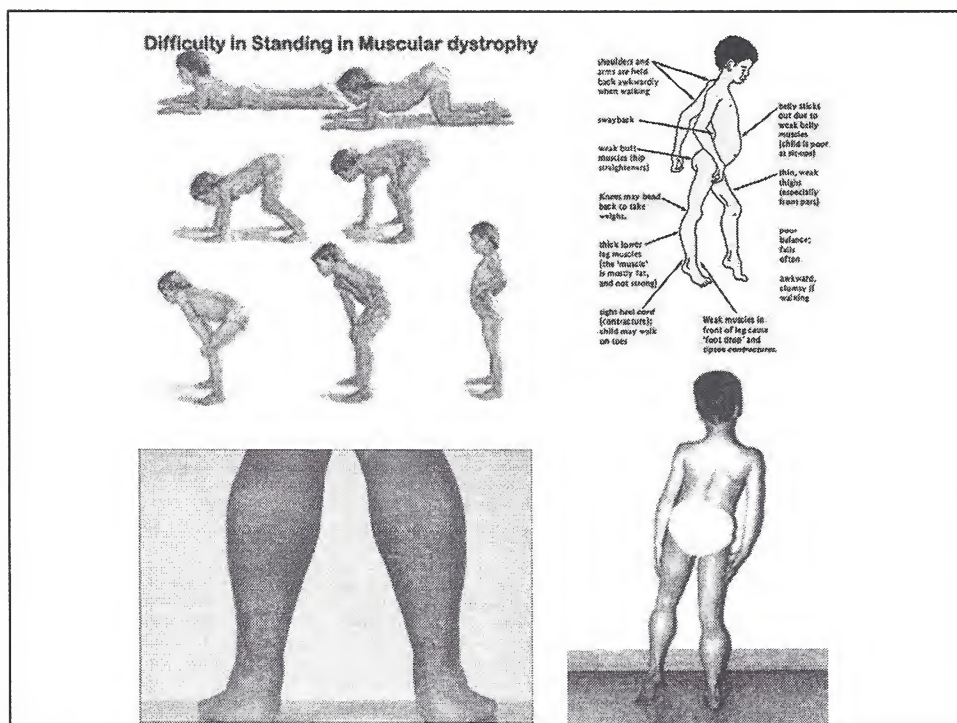
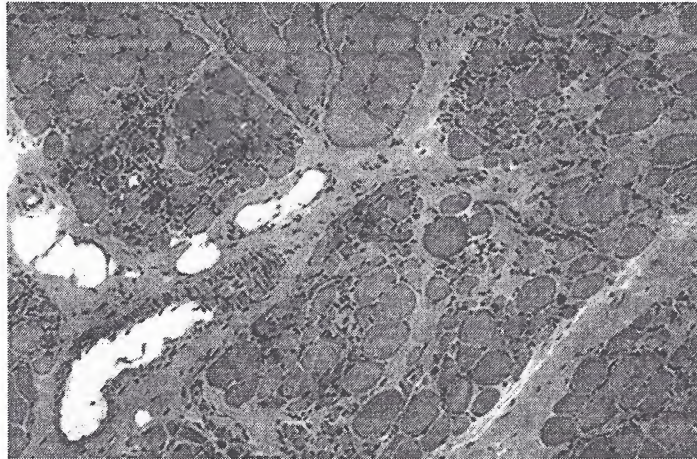


MALATTIE MUSCOLARI EZIOLOGIA

Forme Geneticamente Determinate

Muscular Dystrophies (MD)

Duchenne MD Becker MD	X-linked; infancy
Limb Girdle MD	AD, AR; 1°-4° decade
Emery-Dreifuss MD	X-linked, AD; childhood, adolescence several variants
Facio-Scapulo-Humeral MD	AD; childhood, early adulthood
Oculo-Pharyngeal MD	AD; late-onset
Myotonic Dystrophy	AD; preschool age 2 forms



MALATTIE MUSCOLARI EZIOLOGIA

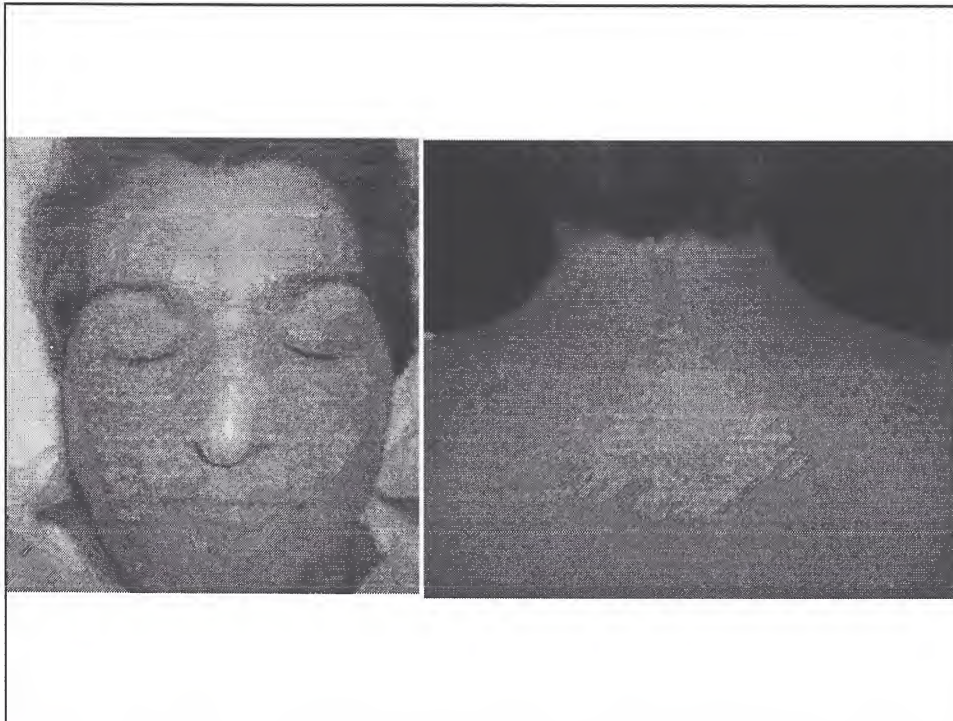
Forme Secondarie

Immuno-mediate	Infiammatoria	Polimiosite Dermatomiosite Miopatia da Corpi Inclusi (IBM)
	Anticorpo-mediata	Miastenia Gravis S miasteniformi
Endocrine	Iper/Ipotiroidismo Iper/Ipoparatiroidismo	
	Ipersteroidismo (sindrome di Cushing)	
Biochimiche	Diabete Mellito Ipokaliemia acquisita Malattie Renali	

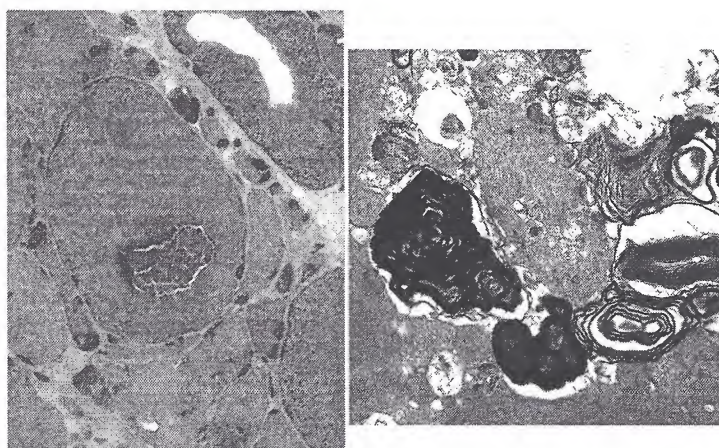
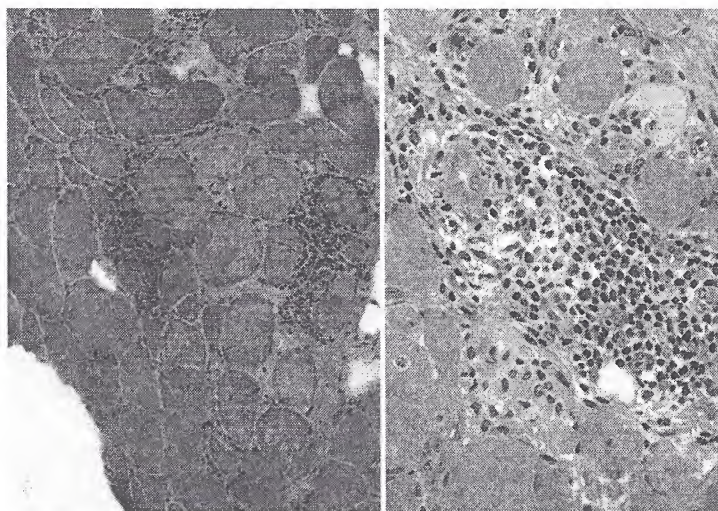
MALATTIE MUSCOLARI EZIOLOGIA

Forme Secondarie Immuno-Mediate

Infiammatoria	Polimiosite Dermatomiosite Miopatia da Corpi Inclusi (IBM)
Anticorpo-mediata	Miastenia Gravis S miasteniformi



	PM	DM	IBM
Age at onset	>18yrs	Adulthood, childhood	>50yrs
sex	M=F	F>M	M>F
Weakness	proximal	proximal	Proximal, early distal involvement
Familial association	No	No	Yes, in some cases / familial inflammatory myopathies /
Response to treatment	good	better	poor
CTDs	yes	yes	Yes, in up to 20%
malignancy	No	yes, in up to 15% of cases	No
Rash	Absent	Present	Absent
Biopsy	"primary" inflammation with the CD8/MHC-I complex & vacuoles	Perifascicular, perymysial, or privascular infiltrates, perifascicular atrophy	Primary inflammation with CD8/MHC-I complex; vacuolated fibers with b-amyloid deposits , cytochrome oxygenase-negative fibers ; signs of chronic myopathy

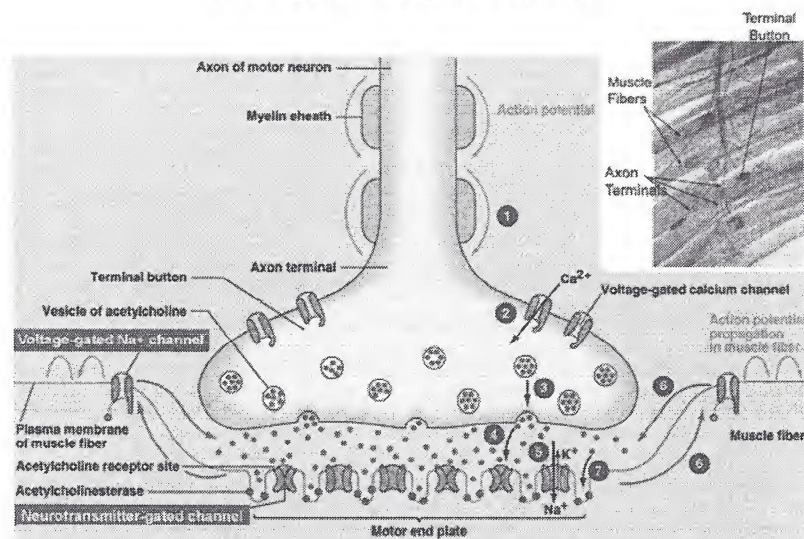


MALATTIE MUSCOLARI EZIOLOGIA

Forme Acquisite

Infettive	virus, parassiti
Tossiche	statine, steroidi, cocaina

The Neuromuscular Junction



MIASTENIA GRAVIS

Clinica

Affaticamento Muscolare (migliora con il riposo)

Tendenza al Reclutamento dei Distretti Muscolari:
coinvolgimento muscolatura facciale: ptosi, disfonia, disfagia



arti, collo.....torace

Diagnostica

Test al Tensilon

EMG (stimolazione ripetitiva)

RMN Torace

Abs anti-Ach-R

Terapia

Farmaci anticolinesterasici

Timectomia

Immunosoppressione: Ig vena

Steroidi

Chemioterapici